Molecular study of CYP21A2 gene for prenatal diagnosis of congenital adrenal hyperplasia. Report of a family Utilidad del estudio molecular de CYP21A2 en el manejo prenatal de hiperplasia suprarrenal congénita: Detección de dos nuevas mutaciones en Chile

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Prenatal treatment of pregnancies at risk of congenital adrenal hyperplasia (CAH) may prevent ambiguous genitalia in female fetuses. We present the prenatal treatment performed in an extended family with two mutations. The proband, a boy with CAH-salt losing form, and his relatives were studied. The proband's paternal uncles/aunts were married to the maternal aunts/uncles, respectively. The relatives had normal basal and stimulated 170HProgesterone levels, which did not clarify their carrier status. The CYP21A2 gene was sequenced. The proband and the paternal relatives harbored a Q318X, R483W mutation in one alíele. The maternal relatives and the proband exhibited an R483 frameshift mutation. Early dexametasone treatment was given during two pregnancies and stopped when male gender was confirmed by early ultrasonography Both newborns were healthy and had normal 170HProgesterone levels. This family had three mutations which abolish the 21-hydroxilase activity. Two mutations were detecte